

Karyotype Report

Customer sample ID: hvs577c
 Internal sample ID: NL63GSAUMCD100020

Gender
 Stated by customer: Male
 According to array data: Male

Copy Number Analysis

Copy number events are reported when they exceed 50 kb in size, have a confidence value greater than 50, and are confirmed by visual inspection of the B-allele frequency (BAF) and log R ratio profiles. CNVs located in known hot spot regions of recurrent mutations in hPSCs are additionally highlighted in the summary table. Loss of heterozygosity (LOH) are reported for regions larger than 1 Mbp.

The reported CNVs can be checked against the [Database of Genomic Variants](#) which provides a comprehensive summary of structural variation in the human genome. The content of the database only includes structural variations identified in healthy control samples.

Affected genes can be monitored by using a genome browser such as the NCBI [Genome Data Viewer](#), [Ensembl Genome Browser](#) or the [UCSC Genome Browser](#).

SampleID	Chr	Start	End	Size	Value	Hotspot region	LOH region
NL63GSAUMCD100020 [17]	3	65797540	67925920	2128380	2	no	yes
NL63GSAUMCD100020 [17]	8	128243131	128372853	129722	3	no	no
NL63GSAUMCD100020 [17]	14	38443309	41906289	3462980	2	yes	yes

Conclusion: The sample shows a duplication on chr 8 as well as LOH regions on chr 3 and 14 with the lengths specified in the table.

Karyogram





Karyotyping

Technical annex

Technology used: Illumina BeadArray

Product: Global Screening Array + Multi Disease content 24
v3 BeadChip

Manifest file: GSAMD-24v3-0-EA_20034606_A1.bpm
Cluster file: GSAMD24v3-0-EA_20034606_A1.egt

Chip barcode and segment: 209838860068 R09C02

Batch ID and 96 well position: WG7941481-MSA3 A03

Call rate: 0.9954867

Typing

Scanner: Illumina iScan, S/N: N263
Site of processing: Life&Brain GENOMICS, Bonn, Germany
Manufacturer: Illumina, Inc., San Diego, United States of America

Genotype Analysis

Genome Studio: GenomeStudio V2.0.5
Genotyping module: Vers. 2.0.5

Copy Number Analysis

Algorithm applied: cnvPartition
Version: 3.2
Software producer: Illumina, Inc., San Diego, United States of America

Analyst

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